E-ISSN 2250-0944
ISSN 2250-1150
doi: 10.31838/ijprt/16.02.568

Research Article

Pattern of Musculoskeletal Congenital Anomalies in Newborns: A Hospital-Based Cross-Sectional Study

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Received: 15.07.2026, Revised: 17.08.2026, Accepted: 19.09.2026

ABSTRACT

Background: Congenital anomalies are important contributors to neonatal morbidity and mortality, with musculoskeletal abnormalities constituting a substantial proportion of structural birth defects. Early recognition facilitates appropriate evaluation, counselling, and timely intervention.

Objective: To determine the pattern of musculoskeletal congenital anomalies among newborns and assess congenital anomalies in relation to relevant maternal and perinatal factors.

Methods: This observational, cross-sectional analytical study included 1,500 newborns delivered at Mahavir Institute of Medical Sciences, Vikarabad from 2023-2025. All newborns were examined for congenital anomalies soon after birth and within 72 hours. Newborns with detected abnormalities underwent detailed clinical evaluation and relevant investigations. Maternal, antenatal, and perinatal factors were recorded.

Results: Congenital anomalies were identified in 148 of 1,500 newborns (9.9%). Musculoskeletal anomalies constituted the most frequently involved system, affecting 48 (32.4%) newborns with congenital anomalies. Among these 48 cases, clubfoot was the most frequent abnormality (22; 45.8%), followed by polydactyly (16; 33.3%) and syndactyly (10; 20.8%).

Conclusion: Musculoskeletal anomalies constituted the predominant group of congenital abnormalities, with clubfoot being the most frequent manifestation. Systematic neonatal examination can facilitate early identification and appropriate management.

Keywords: Congenital Anomalies, Musculoskeletal Anomalies, Newborn, Clubfoot, Polydactyly, Syndactyly.

INTRODUCTION

Congenital anomalies are structural, functional, or metabolic abnormalities that originate during intrauterine life and may be identified prenatally, at birth, or later in infancy. They result from disturbances in embryogenesis or developmental processes and vary considerably in their clinical severity, ranging from minor morphological abnormalities to major defects associated with substantial morbidity, disability, or mortality.[1,2] Congenital anomalies can involve virtually any organ system, and their clinical implications depend on the type, extent, and associated abnormalities.

Congenital anomalies represent an important global health concern because of their contribution to neonatal and childhood mortality and long-term disability. The burden is particularly relevant in low- and middle-income countries, where limitations in antenatal screening, nutritional deficiencies, maternal infections, environmental exposures, and restricted access to specialized care may influence both occurrence and outcome.[3] In India, the reported prevalence of congenital anomalies varies considerably across populations and healthcare settings. Differences in study design, inclusion criteria, diagnostic facilities, timing of examination, and referral patterns may partly account for this variation.[4,5]

The etiology of congenital anomalies is heterogeneous and often remains unidentified. Recognized determinants include genetic and chromosomal abnormalities, environmental teratogens, maternal infections, nutritional deficiencies, and maternal medical disorders.[1,6] Consanguinity has been associated with an increased occurrence of uncommon genetic disorders and congenital

abnormalities, whereas advanced maternal age is an established risk factor for chromosomal abnormalities. Maternal diabetes, hypertension, exposure to potentially teratogenic substances, inadequate folate intake, and certain infections during pregnancy may also influence fetal development.[6,7]

Musculoskeletal congenital anomalies constitute an important component of the overall spectrum of birth defects. They encompass abnormalities affecting the limbs, bones, joints, and related structures and may occur as isolated defects or as components of multisystem anomalies and syndromes. Common clinically recognizable abnormalities include congenital talipes equinovarus (clubfoot), polydactyly, and syndactyly. Their spectrum varies across populations, and several Indian hospital-based studies have reported the musculoskeletal system among the most frequently affected organ systems in newborns with congenital anomalies.[5,8]

Identification of musculoskeletal anomalies immediately after birth is particularly important because many are externally visible and can be detected through a systematic head-to-toe neonatal examination. Early recognition enables assessment for associated abnormalities and facilitates appropriate referral, counselling, and management. Some musculoskeletal abnormalities can be managed effectively when treatment is initiated early, thereby reducing subsequent functional disability. Newborn screening and detailed physical examination therefore represent important components of congenital anomaly detection.[3,9]

MATERIALS AND METHODS

Study Design and Setting

This was an observational, cross-sectional, analytical study conducted in Mahavir Institute of Medical Sciences, Vikarabad. The study population comprised newborns delivered at the study institution.

Study Population and Sample Size

A total of 1,500 newborns were included in the study. The sample size estimation was based on a previous study by Sarkar et al., which reported a prevalence of congenital malformations of 2.22%. Sample size calculation was performed using WINPEPI software at a 95% confidence level. The final sample size was rounded up to 1,500 newborns.

Eligibility and Newborn Screening

All newborns delivered at Mahavir Institute of Medical Sciences, Vikarabad, during the study period from 2023-2025 were eligible for evaluation. Each newborn was examined for congenital anomalies soon after birth and within the first 72 hours of life. Newborns in whom an anomaly was identified either antenatally or during postnatal examination underwent detailed assessment according to the predefined study proforma.

For the present manuscript, the analysis focuses specifically on musculoskeletal congenital anomalies identified within the overall cohort of newborns screened for congenital abnormalities.

Data Collection

Information was collected regarding maternal sociodemographic characteristics, antenatal and obstetric history, and relevant perinatal factors. The variables documented in the original study included socioeconomic status, consanguinity, parity and birth order, first-trimester febrile illness, previous abortions, infertility treatment, sibling deaths and congenital anomalies among siblings, smoking and alcohol exposure, polyhydramnios or oligohydramnios, and maternal medical conditions.

Socioeconomic status was assessed using the revised B. G. Prasad socioeconomic classification. Maternal antenatal risk factors documented in the study included hypothyroidism, pregnancy-induced hypertension, gestational diabetes mellitus, TORCH infection, hyperthyroidism, bronchial asthma, hepatitis B, HIV, seizures, antiphospholipid antibody positivity, idiopathic thrombocytopenic purpura, and Sjögren syndrome.

Perinatal Assessment

Perinatal variables included gestational age, birth weight, sex of the newborn, and mode of delivery. Newborns were categorized as preterm or term according to gestational age. Birth weight was recorded and categorized into <1 kg, 1–2.5 kg, 2.5–3.5 kg, and >3.5 kg for analysis in the original study. Mode of delivery was classified as lower-segment caesarean section or normal vaginal delivery.

Clinical Examination for Congenital Anomalies

A systematic head-to-toe examination was performed to identify congenital abnormalities. The assessment included examination of the head and face, eyes, nose, ears, mouth,

tongue, palate, jaw, neck, back, genitalia, skin, and upper and lower extremities. Particular attention was given to externally recognizable musculoskeletal abnormalities.

The musculoskeletal anomalies documented in the dedicated musculoskeletal analysis were:

- Congenital talipes equinovarus (clubfoot)
- Polydactyly
- Syndactyly

The thesis also recorded developmental dysplasia of the hip during the broader head-to-toe examination; however, the dedicated musculoskeletal results table used for the primary subtype analysis contained 48 cases comprising clubfoot, polydactyly, and syndactyly. Therefore, the present manuscript retains the dedicated Results table as the primary source for subtype frequencies rather than attempting to reconcile differing counts reported elsewhere in the thesis.

Classification of Congenital Anomalies

Congenital anomalies were categorized as major or minor according to their clinical significance in the original study. A major anomaly was defined as an abnormality adversely affecting health, functioning, or social acceptability, particularly abnormalities involving major organs such as the heart, kidneys, or brain. A minor anomaly was considered to have limited medical or social significance.

Among the 148 newborns with congenital abnormalities, 124 (83.8%) had major anomalies and 24 (16.2%) had minor anomalies.

Diagnostic Evaluation

When congenital anomalies were suspected clinically or had been identified during antenatal ultrasonography, further investigations were undertaken as clinically required. These included two-dimensional echocardiography, ultrasonography of the abdomen and pelvis, ultrasonography of the cranium, ultrasonography of the sacral region, infantogram, and magnetic resonance imaging where indicated.

Newborns with congenital anomalies requiring urgent intervention were referred for further specialist management.

Outcome Measures

The primary outcome for the present manuscript was the pattern and relative frequency of musculoskeletal congenital anomalies among newborns with congenital abnormalities.

The principal musculoskeletal outcomes were the frequencies and proportions of clubfoot, polydactyly, and syndactyly. Musculoskeletal involvement was also compared descriptively with other organ-system categories identified in the original study, including cardiovascular, gastrointestinal, genitourinary, respiratory, and central nervous system abnormalities.

The original study additionally evaluated associations between congenital anomalies and maternal/perinatal characteristics, including consanguinity, gestational age, parity, birth weight, and presence of perinatal risk factors.

Statistical Analysis

Data were coded and entered into Microsoft Excel, and statistical analysis was performed using SPSS. Categorical variables were summarized using frequencies and percentages. The chi-square test and Fisher's exact test, where appropriate, were used to assess associations between categorical variables.

Odds ratios were reported for selected risk-factor analyses in the original results. A p-value <0.05 was considered statistically significant.

In the original dataset, statistical significance was demonstrated for the association of congenital anomalies with consanguinity ($p<0.0001$), birth weight <1 kg (OR 4.3; $p=0.019$), and the overall presence of recorded perinatal risk factors ($p=0.022$). Gestational age and parity did not demonstrate statistically significant associations in the reported analyses.

RESULTS

Table 1. Overall Prevalence and Classification of Congenital Anomalies among Screened Newborns

Characteristic	Frequency (n)	Percentage (%)
Total newborns screened	1500	100.0
Congenital anomalies present	148	9.9
Congenital anomalies absent	1352	90.1
Classification among affected newborns (n=148)		
Major anomalies	124	83.8

Minor anomalies	24	16.2

Table 2. Selected Maternal and Perinatal Characteristics Associated with Congenital Anomalies

Factor	Newborns with anomaly, n/N (%)	Statistical finding
Consanguineous marriage	28/147 (19.0)	p<0.0001
Non-consanguineous marriage	120/1353 (8.8)	
Preterm	19/283 (6.7)	p=0.349
Term	129/1217 (10.6)	
Birth weight <1 kg	4/12 (33.3)	OR=4.3; p=0.019
Perinatal risk factors present	21/112 (18.8)	p=0.022
Perinatal risk factors absent	127/1388 (9.1)	

Table 3. System-Wise Distribution of Congenital Anomalies among Affected Newborns (N=148)

System involved	Number of newborns	Percentage (%)
Musculoskeletal system	48	32.4
Gastrointestinal system	36	26.4*
Genitourinary system	34	22.9
Cardiovascular system	28	18.9
Central nervous system	8	9.5*
Respiratory system	3	2.0

Table 4. Pattern of Musculoskeletal Congenital Anomalies (N=48)

Musculoskeletal anomaly	Frequency (n)	Percentage (%)
Congenital talipes equinovarus (clubfoot)	22	45.8
Polydactyly	16	33.3
Syndactyly	10	20.8
Total	**48**	**100.0**

Table 5. Key Findings Relevant To Musculoskeletal Congenital Anomalies

Study parameter	Finding
Total newborns screened	1500
Newborns with any congenital anomaly	148 (9.9%)
Newborns with musculoskeletal involvement	48/148 (32.4%)
Most frequently involved system	Musculoskeletal
Most common musculoskeletal anomaly	Clubfoot, 22/48 (45.8%)
Second most common	Polydactyly, 16/48 (33.3%)
Third most common	Syndactyly, 10/48 (20.8%)

DISCUSSION

The present hospital-based cross-sectional study evaluated congenital anomalies among 1,500 newborns, with particular emphasis on the musculoskeletal system. Congenital anomalies were detected in 148 newborns, corresponding to an overall prevalence of 9.9%. Musculoskeletal anomalies constituted the most frequently involved system, accounting for 48 (32.4%) affected newborns. Within the dedicated musculoskeletal dataset, congenital talipes equinovarus (clubfoot) was the most common abnormality, followed by polydactyly and syndactyly.

The predominance of musculoskeletal anomalies observed in the present study is consistent with findings from several Indian studies. Previous hospital-based studies from eastern and southern India have similarly identified the musculoskeletal system as one of the most frequently affected systems among newborns with congenital anomalies.[5,8] Studies conducted in rural Maharashtra and other hospital-based populations have also demonstrated a substantial contribution of musculoskeletal abnormalities to the overall congenital anomaly burden.[10,11] Variations in the predominant organ system across different studies may be related to differences in study populations, referral patterns, diagnostic facilities, inclusion criteria, and methods used for the detection and classification of congenital anomalies.

In contrast, the predominant system has not been uniform across different study settings. Some studies have reported cardiovascular

anomalies as a major category,[12] whereas others have documented a greater contribution of central nervous system abnormalities.[13] In tertiary paediatric surgical settings, gastrointestinal anomalies have also been reported as an important group of congenital abnormalities requiring intervention.[14] These variations indicate that the observed spectrum of congenital anomalies is influenced considerably by the characteristics of the study population, type of healthcare facility, referral practices, and availability of diagnostic services. Among the 48 musculoskeletal cases included in the dedicated analysis, clubfoot was the most frequent anomaly, affecting 22 (45.8%) newborns, followed by polydactyly in 16 (33.3%) and syndactyly in 10 (20.8%). This distribution is comparable to previous Indian observations in which congenital talipes equinovarus was reported as the most frequent musculoskeletal abnormality, followed by polydactyly and syndactyly.[5] These conditions are generally externally recognizable and can therefore be detected through a careful and systematic physical examination of the newborn. Early recognition is particularly important because it enables timely evaluation for associated abnormalities and appropriate referral for further management.

The present study also identified significant associations between congenital anomalies overall and selected maternal and perinatal characteristics. Congenital anomalies occurred in 19.0% of newborns from consanguineous unions compared with 8.8% from non-consanguineous unions ($p<0.0001$). Birth weight below 1 kg was associated with an increased risk of congenital anomalies (OR=4.3; $p=0.019$). Furthermore, congenital anomalies were significantly more frequent among newborns with recorded perinatal risk factors ($p=0.022$). Previous hospital-based investigations have likewise demonstrated associations between congenital malformations and various maternal and fetal characteristics.[15] These observations support the importance of careful antenatal risk assessment together with systematic examination of newborns.

An important strength of the present study was the systematic examination of newborns soon after birth and within the first 72 hours of life, supplemented by targeted diagnostic investigations whenever indicated. This approach facilitated the identification of externally evident musculoskeletal abnormalities as well as assessment for associated congenital defects. Early detection of clubfoot and digital anomalies is clinically important because it allows timely specialist referral, further evaluation, parental counselling, and appropriate management, potentially reducing subsequent functional impairment.

CONCLUSION

Musculoskeletal anomalies constituted the most frequently involved system (32.4%) among newborns with congenital anomalies in this hospital-based cohort. Congenital talipes equinovarus (clubfoot) was the predominant musculoskeletal abnormality, followed by polydactyly and syndactyly. Systematic examination of newborns during the early postnatal period is important for prompt recognition of these abnormalities and appropriate evaluation and referral. The observed associations of congenital anomalies overall with consanguinity, extremely low birth weight, and recorded perinatal risk factors further support careful antenatal risk assessment and comprehensive neonatal screening.

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